



## L1CAM gene

L1 cell adhesion molecule

### Normal Function

The *L1CAM* gene provides instructions for producing the L1 protein, which is found throughout the nervous system on the surface of nerve cells (neurons). The L1 protein spans the cell membrane, so that one end of the protein remains inside the cell and the other end projects from the outer surface of the cell. This positioning allows the L1 protein to help activate various chemical signals within the cell. The L1 protein plays a role in the development and organization of neurons, the formation of the protective sheath (myelin) that surrounds certain neurons, and the formation of junctions between nerve cells (synapses), where cell-to-cell communication occurs.

### Health Conditions Related to Genetic Changes

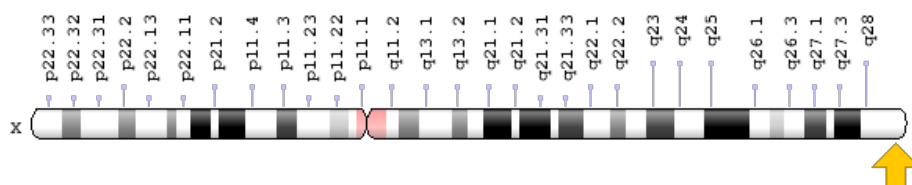
#### L1 syndrome

There are about 200 mutations in the *L1CAM* gene that cause L1 syndrome. These mutations change the structure of the L1 protein or disrupt its production. Mutations that change one protein building block (amino acid) typically cause less severe signs and symptoms than mutations that disrupt the production of the L1 protein. All mutations, however, impair L1 protein function and interfere with the development and function of neurons, leading to the characteristic features of L1 syndrome.

### Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 153,861,514 to 153,886,174 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- antigen identified by monoclonal antibody R1
- CAML1
- CD171
- HSAS
- HSAS1
- L1CAM\_HUMAN
- MASA
- MIC5
- N-CAML1
- neural cell adhesion molecule L1
- S10
- SPG1

## Additional Information & Resources

### Educational Resources

- Genetics for Surgeons (2005, first edition): Hydrocephalus  
<https://www.ncbi.nlm.nih.gov/books/NBK7540/#A593>
- L1CAM Mutation Web Page  
<http://www.l1cammutationdatabase.info/>

### GeneReviews

- L1 Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1484>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28L1CAM%5BTIAB%5D%29+OR+%28L1+cell+adhesion+molecule%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- L1 CELL ADHESION MOLECULE  
<http://omim.org/entry/308840>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/L1CAMID44110chXq28.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=L1CAM%5Bgene%5D>
- HGNC Gene Family: CD molecules  
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Family: Fibronectin type III domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/555>
- HGNC Gene Family: I-set domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/593>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6470](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6470)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3897>
- UniProt  
<http://www.uniprot.org/uniprot/P32004>

## **Sources for This Summary**

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